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Hereditary Cancer Model of Care

Prepared by – NCCP

DRAFT FOR CONSULTATION

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# Forward

To be added post consultation

# Purpose

This document has been developed on behalf of the NCCP Advisory Group on Hereditary Cancer (see Appendix X for membership). Its purpose is to inform development of services for the identification and management of people with an inherited cancer predisposition.

# Abbreviations

|  |  |
| --- | --- |
| **Abbreviation** | **Definition** |
| ANP | Advanced Nurse Practitioner |
| AYA | Adolescents and Young Adults |
| CAYA | Children, Adolescents and Young Adults |
| CHI | Children’s Health Ireland |
| CNS | Clinical Nurse Specialist |
| DNA | Deoxyribonucleic acid |
| DOH | Department of Health |
| EHR | Electronic Health Record |
| ERN | European Reference Network |
| EU | European Union |
| GDPR | General Data Protection Regulation |
| GP | General Practitioner |
| HSE | Health Service Executive |
| ICS | Irish Cancer Society |
| IT | Information Technology |
| KPI | Key Performance Indicators |
| MDT | Multi-Disciplinary Team |
| NCCP | National Cancer Control Programme |
| NCRI | National Cancer Registry Ireland |
| NGS | Next Generation Sequencing |
| NHS | National Health Service |
| NICE | National Institute for Health and Care Excellence |
| PPI | Public and Patient Involvement |
| PPPGs | Policy, Procedures, Protocols and Guidelines |
| SJH | St. James’s Hospital |
| UKCGG | UK Cancer Genetics Group |
| WGS | Whole Genome Sequencing |
| WHO | World Health Organisation |

# Glossary

|  |  |
| --- | --- |
| **Name** | **Definition** |
| Carrier | A person carrying a pathogenic variant of a gene. For many cancer predisposition syndromes, one copy of the gene is enough to increase their risk of developing cancer. For other conditions, including some less common cancer predisposition syndromes, two copies of the gene are required. In this case, a carrier is not themselves at risk of the syndrome but they may still pass on the gene conferring that risk to their children. |
| Cancer predisposition | Having a predictable increased risk of developing cancer compared to the general population, e.g. due to inheriting a pathogenic gene variant (germline mutation) |
| Diagnostic testing | See *genetic testing* |
| Gene | The basic physical and functional unit of heredity. Genes, which are made up of DNA, act as instructions to usually make molecules called proteins. |
| Genetic testing | **Germline genetic testing** tests for the presence of germline genetic variants - A germline genetic variant (or germline mutation) is a gene change that is passed down from parent to offspring, then incorporated into the DNA of every cell in the body of the offspring. Germline testing is usually carried out on blood or saliva. Germline gene variants are also known as hereditary gene variants.  **Somatic genetic testing** tests for the presence of somatic mutations – A somatic mutation is an alteration in DNA that is not inherited. It may arise sporadically or environmental/ lifestyle factors may induce a somatic change. Somatic mutations can occur in any cell of the body except for germ cells and therefore are not passed on to any offspring. These alterations can (but do not always) cause cancer or other diseases.  **Diagnostic testing** is testing offered to symptomatic people to determine whether they have a condition, e.g. a cancer predisposition syndrome, associated with the symptoms they already have, such as an existing personal diagnosis of cancer.  **Predictive testing** is used to look for a pathogenic gene variant in people who have no symptoms of the associated disease/condition, such as cascade testing of a blood relative for a pathogenic variant which has been identified in an affected family member |
| Genetics | Genetics – the study of genes, genetic variation, and heredity in living organisms |
| Genomics | Genomics - The application of genome-based knowledge through the study of genes and other genetic information, their functions and interrelationships for the benefit of human health. |
| Germline genetic testing | See *genetic testing* |
| Hereditary cancer services | Referring to the service in its entirety, from appropriate identification of people who may have an inherited cancer predisposition (germline genetic variant/mutation), through assessment and testing, and appropriate clinical management and support to maximise health and wellbeing. |
| Mainstreamed testing/ mainstreaming | Diagnostic genetic testing for patients with a diagnosis of cancer, outside of genetics services and as part of standardised cancer care and in line with nationally agreed criteria and protocols, including appropriate education and training. |
| Pathogenic variant | An alteration in a gene, which leads to an increased risk of disease, e.g. cancer. Used in preference over the term gene ‘mutation’ |
| Predictive testing | See *genetic testing* |
| Somatic testing | See *genetic testing* |
| Variant of Uncertain Significance | A Variant of Uncertain Significance (VUS) is a genetic change that the laboratory cannot interpret as either harmless (benign) or potentially disease-causing (pathogenic), as the meaning of this type of genetic change is not yet known |

# Executive Summary

Genetics is now recognised as a growing and integral part of cancer care, with a role across treatment, early detection and risk reduction. Identification of cancer predisposition will only lead to patient benefit, if necessary services are in place and appropriately resourced and monitored. The potential benefit to a patient - and their relatives - can include reducing the risk of cancer developing, identifying cancer at an earlier more treatable stage, informing the optimal choice of targeted therapy if a cancer develops and facilitating reproductive options for the individual.

* **Risk reduction** – Genetics offers a direct and cost-effective way of identifying people who are at high risk of developing cancer. These people can then be managed appropriately, including offering interventions to reduce the risk of cancer developing, e.g. risk-reducing surgery and chemoprevention.
* **Early detection** – People who are identified as having an inherited cancer predisposition can be closely monitored to facilitate early detection and treatment of disease. Diagnosing cancer early typically allows for less complex treatment and optimal outcomes.

* **Treatment** - Certain cancer therapies rely on genetic and genomic information to tailor treatment to give the best chance of cure. For cancer patients with an inherited cancer predisposition, treatment decision-making based on timely genetic testing is rapidly becoming an international standard of care. Genetic test results guide surgical decisions and direct choice of medication. Targeting medication according to genetic test results (pharmacogenetics) is at the forefront of precision medicine.
* **Reproductive counselling and care** – Additional benefits of cancer genetics include the ability to identify people with a cancer predisposition and offer them appropriate counselling and care in relation to their reproductive health and options.
* **Benefit to relatives** – Cascade or predictive testing of unaffected relatives identifies more individuals with an inherited cancer predisposition, who can then benefit from the above cancer risk reduction and early detection measures.

The identification of cancer predisposition allows for the implementation of these risk-reduction measures, such as prophylactic surgery and chemoprevention, and surveillance to ensure early detection. In this way, genetic testing - for individuals and subsequent cascade testing of relatives - and provision of comprehensive downstream services can effectively reduce both cancer incidence and cancer mortality.

The **vision of this model of care** is that it will provide clarity regarding the structure and governance of hereditary cancer services in Ireland. The model of care aims to ensure:

* Development of national guidance to underpin standardised best practice, including mainstreaming of cancer genetic testing
* Equitable access to hereditary cancer services for all
* Clear governance and clarity regarding the roles of national genetics and genomic structures, specialist cancer genetics services and regional cancer predisposition services, within an overarching model of care
* Commitment to the Sláintecare principles of right person, right place, right time – through the integration of genetic assessment and testing into regional services, with access to national specialist genetic expertise as required
* Appropriate use of technology
* Timely access to genetics assessment and testing, to specialist cancer genetics expertise as required and to risk reduction options
* Comprehensive and coordinated approach to ongoing management
* Quality assurance of service
* Access to high quality patient information, psychological support and peer support for patients
* Education and training of health care professionals, including accredited training for those delivering mainstreamed pathways

## Three tier model of service delivery

A programmatic approach is required to ensure national agreement of guidance, including mainstreaming protocols, laboratory and clinical data infrastructure, specialist genetics workforce development, and education of the wider workforce. This programme will be a collaboration between by the National Genetics and Genomics Office and the National Cancer Control Programme.

Delivery of hereditary cancer services will be organised across three ‘tiers’:

* Generalist level
* Regional Cancer Predisposition Services
* Specialist Cancer Genetics Services

This model of care outlines roles of each tier and the working relationships to be established between the ‘tiers’, e.g. the way in which the Specialist Cancer Genetics Service will support and enable delivery of care by Regional Cancer Predisposition Services. It is essential that patient care across the tiers is seamless, with clear communication and coordination of care.

**Figure 1: Hereditary cancer services - model of service delivery**



**Generalist Services**

Healthcare services (including primary care and non-oncology services in secondary care) should be empowered to recognise those with a possible inherited predisposition to cancer and refer appropriately and to understand the management of those with identified cancer predisposition.

Referral pathways and protocols will be agreed nationally, and specific education initiatives should focus on those working in generalist services, to ensure appropriate knowledge of referral pathways/protocols and identification of patients for referral. The need for improved genetic literacy across all healthcare professionals is recognised in the draft National Genetics and Genomics Strategy.

## Regional Cancer Predisposition Services

Regional services have a role in the identification and assessment of those with a possible inherited predisposition to cancer, including [mainstreamed diagnostic testing](#_The_concept_of) for inherited pathogenic gene variants in patients with cancer, according to national protocols.

Regional services also deliver the necessary healthcare to those identified with an inherited cancer predisposition, to determine role for targeted cancer therapy where relevant, to reduce cancer risk and/or facilitate early detection and to maximise patient health and wellbeing.

Multiple services including medical oncology, surgical oncology, haematology and benign services such as gastroenterology are involved in the identification and management of cancer predisposition.

The required multi-disciplinary service for people with the more common cancer predisposition syndromes (e.g. BRCA carriers, Lynch syndrome) should be coordinated regionally at cancer centre level and requires a structure to facilitate follow-up of patients. Certain rarer syndromes may follow a different model, with patient management and follow-up delivered in one centre, in close collaboration with the specialist cancer genetics service.

Regional cancer predisposition services require designated input from specialist cancer genetics services, with a clear clinical governance structure.

### Regional Cancer Predisposition Services - Workforce

#### **Clinical Lead Role**

A **Clinical Lead for Cancer Predisposition** should be appointed with an identified sessional commitment to lead the development of each Regional Cancer Predisposition Service. This role will include leading:

* development of regional service (tailored to population size and specialisms)
* adherence to national protocols
* development of local pathways
* ensuring appropriate education and training of local workforce. All cancer services should be upskilled to ensure the provision of a high-quality service capable of accurately assessing family history risk (in patients affected/ unaffected by cancer) and to effectively implement mainstreamed testing pathways (related to drug treatment or other indications for treatment).
* clinical audit & service improvement
* service monitoring, including measuring and reporting KPIs

***Requirement:*** *Each regional service will require an identified clinical lead for this role, e.g. with a sessional commitment equivalent to 0.4WTE. A lesser time commitment may be feasible in smaller centres, or if sharing this role across two sites within a network/ future Regional Health Area.*

#### **Administrative support**

Appropriate **administrative support** will be integral to service delivery, monitoring and evaluation. These roles should include data management and clinic coordination. Genetics and genomics has a higher requirement for record retrieval and documentation than other specialties, due e.g. to the detailed information provided by patients on their family history, or the tracking of multiple pathology test results. Genomics Resource Associates can play an essential non-clinical support role in this area, including support for patients in navigating the assessment and testing process and the subsequent management.

Technological solutions may alleviate some of administrative burden associated with service delivery.

#### **Genetic Counsellors**

The role of **Genetic Counsellor** is a Specialist Cancer Genetics role, but will be delivered regionally. It will be possible for a Genetic Counsellor to be either appointed and employed locally by the Regional Cancer Predisposition Service, or centrally by the Specialist Cancer Genetics Service. Where appointed regionally, a professional reporting relationship to and sessional commitment to Specialist Cancer Genetics will be required.

A Genetic Counsellor will fulfil the following roles within each of the Regional Cancer Predisposition Services:

* act as point of contact for support for non-geneticists who are providing pre-test counselling
* develop and deliver educational initiatives to the wider workforce
* provide comprehensive pre or post-test genetic counselling as required, including reproductive advice/information
* act as point of contact/link to the Specialist Cancer Genetics tertiary service for complex cases and consultation on variants of unknown significance
* contribute to research

***Requirement:*** *Each of the regional cancer predisposition services requires an identified genetic counsellor, appointed either locally or centrally, with a professional reporting relationship to a Principal Genetic Counsellor in the specialist cancer genetics service.*

*This will not equate to a WTE cancer genetics counsellor per cancer centre, as a sessional commitment will be included to the national hub/network, including national MDTs.*

*Smaller cancer centres may choose to operate as part of a network (/future Regional Health Area) with another cancer centre for this service.*

#### **Specialty Lead**

Within specific specialties (e.g. medical oncology, colorectal, gynaecology, breast), a **lead consultant or ANP** should lead on implementing Regional Cancer Predisposition Services, including ensuring access to mainstreamed testing where applicable and ongoing management of those with an identified cancer predisposition.

The exact configuration of specialties and their role in delivery of Regional Cancer Predisposition Services will vary according to services available in the region and may change over time – e.g. breast, gynaeoncology, colorectal, upper GI, medical oncology, haematology. Collaborative working will be key, as patient needs frequently cross clinical specialties.

#### **Nursing**

CNS and ANP members of teams are key to the delivery of the Regional Cancer Predisposition Services, with roles including assessment of family history, patient pre-test counselling and provision of continuity of care and coordinated care for those identified with a cancer predisposition. Administrative support in this area will allow skilled nursing staff to work at the top of their licence and will support the coordination of care across different clinical areas.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Sample staff in Regional Cancer Predisposition Service** | | | | | |
|  | | | | **Clinical Lead** |  |
| **Medical Oncology** | **Colorectal**  **service** | **Gynaecology** | **Breast service** |  | **Genetic Counsellor\*** |
| *Consultant* | *Consultant* | *Consultant* | *Consultant* | Admin staff |  |
| *CNS* | *CNS* | *CNS* | *CNS* | Genetics Resource Associate | |
| *ANP* | *ANP* | *ANP* | *ANP* |  |  |
| *Pathology, radiology, endoscopy, psychooncology services* | | | | | |

\*may be employed by cancer centre or specialist cancer genetics service but professional reporting relationship to specialist cancer genetics

## Specialist Cancer Genetics Service

The Specialist Cancer Genetics Service will function as the national centre for clinical expertise in Ireland and will provide support and advice to Regional Cancer Predisposition Service sites via a network of designated genetic counsellors, a tertiary referral service and national MDT(s).

The role of the Specialist Cancer Genetics Service will include:

* develop national guidance, including protocols for mainstreamed services as appropriate. All standards, protocols and guidelines will be agreed nationally, and founded on the best available evidence and best international practice, appropriately applied in the Irish context.
* horizon scanning - as cancer genetics and genomic services and techniques continue to advance, it is important to ensure that the introduction of new tests and phasing out of others is informed by specific guidelines, and that this will be audited;
* develop and deliver educational initiatives to the wider workforce. Guidance is specifically required to support the implementation of mainstreamed pathways.
* support non-geneticists who are providing pre-test counselling
* provide a regional predictive genetic testing service
* provide comprehensive pre or post-test genetic counselling as required, including reproductive advice/information
* provide a tertiary referral service for complex cases and consultation on variants of unknown significance (VUS) – referral pathways will be agreed to facilitate referral of patients to the Specialist Cancer Genetics Service by a clinical team based at the Regional Cancer Predisposition Services.
* Support in the management of complex cases may be provided remotely or in person at a tertiary referral centre (+/- discussion at a national MDT).

Adult and paediatric patients have differing needs and should be assessed and managed in an appropriate environment (see below).

### **Specialist Cancer Genetics Services - Workforce**

**Consultants**

Consultant expertise in specialist cancer genetics is provided by (i) those trained as Consultants in Clinical Genetics and (ii) those trained as oncology consultants, who have developed expertise in the area of cancer genetics, specifically cancer predisposition. Those trained in oncology have a greater understanding of the overlap with current or future cancer treatments, whereas clinical geneticists have a broader role in areas such as syndrome overlap with non-cancer areas. Both areas of expertise are valued and should be maintained.

Of note, the oncology consultant role referred to here is different to that defined within Regional Cancer Predisposition Services. The lead role regionally may be held by someone with an interest in service development in this area but would not require the clinical expertise to provide a tertiary referral specialist cancer genetics service. However, regional expertise can be expected to develop further over time, particularly in the area of more common hereditary cancer syndromes.

**Genetic Counsellors**

As previously outlined, some genetic counsellors will provide a dedicated service to regional cancer predisposition services but will maintain both a sessional commitment and a professional reporting relationship to the specialist cancer genetics service.

Genetic Counsellors will also operate as part of the tertiary referral service in cancer specialist genetics, will have a role in education, training and guidance development and provide a clinical governance structure for genetic counsellors working regionally.

Challenges in Ireland have been noted in relation to lack of career pathway and registration for genetic counsellors. This has been highlighted as a priority issue for the national genetics and genomics office.

**Administrative and IT support**

As outlined for regional services, the administrative workload for genetics is high compared to other specialties. The team will also benefit from Genetic Resources Associates and support to ensure coordination of national MDT(s) and the genetic counsellor network.

**Laboratory expertise**

Molecular diagnostic expertise is required as part of the specialist cancer genetics MDT, particularly in relation to the interpretation of Variants of Uncertain Significance.

## Paediatric and AYA Cancer Predisposition Service

Childhood cancer, including that in adolescents/young adulthood, is a rare but complex disease, with differing cancers and different disease biologies to those seen in adults. In addition, these cancers are associated with a higher rate of cancer predisposition, with less influence of environmental factors. Currently, it is estimated that up to 30% of young patients with childhood or adolescent/young adult cancers (CAYA) require genetic evaluation, with at least 10% having a confirmed cancer predisposition syndrome.

A comprehensive service is therefore required that incorporates cancer predisposition risk assessment for all CAYA patients. In addition, as standard of care is moving internationally towards somatic testing of all tumours diagnosed in the CAYA population, germline testing for all will likely be required, either in sequence or parallel, necessitating expertise in relation to interpretation and management.

Children’s cancer services are already centralised at the CHI cancer centre (currently in Crumlin) and the development of a formal national network for CAYA services is currently underway (reference). The provision of a single national specialist paediatric MDT for cancer genetics fits with this model and such a service has already commenced at CHI at Crumlin. The multidisciplinary input required crosses specialist genetics, oncology/haematology, molecular pathology and the wider CAYA cancer team. Collaboration will be required between this MDT and that at the AYA service in St James’ Hospital, to meet the needs of this age group.

In addition to providing access from regional paediatrics services to/from this specialist service, consideration should also be given to cross over with regional cancer predisposition services who are dealing with adults. For example, it may be appropriate for genetic testing to be carried out in children of adults diagnosed with certain cancer predisposition syndromes. These children should be referred to specialist genetics services associated with paediatric cancer predisposition services, for age-appropriate genetic counselling and management. The benefit of consultant clinical geneticists and genetic counsellors on the MDT is that they are skilled to deal with all ages and all types of clinical presentations (syndromic/non-syndromic), cancer risks and risks of other medical conditions, across all age spectrums.

Similarly, when cancer predisposition is identified in a child, testing of adults may identify those with the same predisposition and who will benefit from ongoing management in their regional cancer predisposition service.

Specific challenges can arise in relation to disease surveillance in this population, particularly younger children. These include greater reliance on U/S and MRI techniques, as minimising use of radiation, and the need for a general anaesthetic for certain surveillance procedures.

## Implementation

The implementation of this Model of Care will be a multi-year project. An implementation group will be established to oversee this work, encompassing the following areas:

|  |
| --- |
| Thematic Area 1: |
| Establish oversight for the implementation of the Hereditary Cancer Model of Care |
| 1.1 Establish a governance and reporting structure aligned to the National Cancer Strategy and National Cancer Control Programme |
| 1.2 Agree areas for collaboration with the National Genetics and Genomics Office once in place |
| 1.3 Convene a multi-disciplinary implementation steering group, with clinical leadership and patient involvement |
| 1.4 Establish relevant working groups for thematic priorities |
| 1.5 Agree an overarching governance framework for hereditary cancer service |
| 1.6 Appoint Clinical Leads for Cancer Predisposition within regional services |
| 1.6 Report at minimum annually on progress in implementation |
| Thematic Area 2 : |
| Development of networked services and MDTs |
| 2.1 Define the process(es) by which genetic counselling services can be networked and made accessible regionally |
| 2.2 Develop a standardised pathway to a national specialist cancer genetics MDT (adults) |
| 2.3 Develop a standardised pathway to a national specialist cancer genetics MDT (CAYA) |
| 2.4 Establish a network of regional cancer predisposition leads |
| Thematic Area 3 : |
| Healthcare professional education and training; workforce development |
| 3.1 Identify the training needs of staff working in generalist settings |
| 3.2 Identify the training needs of staff working in regional cancer predisposition services |
| 3.3 Develop an accredited online programme to meet the minimal learning needs staff working in regional services |
| 3.4 Identify or develop further training courses to meet identified needs of staff |
| 3.5 Work with the National Genetics and Genomics Office, professional training bodies and regulators to develop career pathways in specialist cancer genetics |
| Thematic Area 4: |
| Clinical guidance and pathway development |
| 4.1 Agree the guidance development and approval process, in line with HSE policy, including guidance on laboratory testing |
| 4.2 Establish guidance development groups for common presentations and cancer predispositions |
| 4.3 Ensure patients are involved in the co-design of clinical pathways |
| 4.4 Agree the process by which international guidance relating to rarer conditions will be approved and disseminated for use in Ireland |
| 4.5 Agree the platform(s) through which current guidance will be made available to all |
| 4.5 Highlight critical points in clinical pathways, which should be subject to regular audit |
| 4.6 Describe resource requirements for implementation of pathways |
| Thematic Area 5: |
| Patient Information and supports |
| 5.1 Establish subgroups for development of patient information, according to common presentations and cancer predispositions |
| 5.2 Ensure patients are engaged in the co-design of patient information materials |
| 5.3 Consider how best to meet the information needs of underserved populations and those with health literacy challenges |
| 5.4 Agree or develop platform(s) through which patient information can be most easily accessed |
| 5.5 Ensure evaluation of information materials and supports provided |
| Thematic Area 6: |
| Data Requirements and IT infrastructure |
| 6.1 Explore the role of virtual patient assessment within hereditary cancer services |
| 6.2 Identify the IT infrastructure required to enable the implementation of this model of care, including a genetic counselling network and referral to a national specialist genetics MDT. |
| 6.3 Examine potential data capture options with the National Cancer Information System |
| 6.4 Engage with the National Genetics and Genomics Office in relation to development of registries relation to cancer predisposition |
| 6.5 Agree minimum datasets for common presentations and cancer predispositions |
| 6.6 Agree key performance indicators for the delivery of specialist cancer genetics and regional cancer predisposition services |

# Introduction

Cancer is a leading cause of morbidity and mortality in Ireland, giving rise to approximately 43,600 new cases and 9,400 deaths annually (NCRI). Cancer is caused by changes in the genes that regulate how cells grow and multiply. These genetic changes can result from:

* random errors that occur in DNA as cells multiply
* damage to cellular DNA arising from exposure to carcinogens (e.g. tobacco smoke, UV rays from the sun, and the human papillomavirus (HPV))
* the presence of a pathogenic gene variant, which confers an increased predisposition to cancer.

The majority of cancer cases (approximately 90%) are ‘sporadic’ and arise as a result of an accumulation of damage to DNA, resulting from the first two processes outlined above – random errors that occur in DNA as cells multiply, and/or DNA damage arising from exposure to carcinogens.

Approximately 5%-10% of cancer cases are associated with the presence of a germline pathogenic gene variant. These are genetic variants that were present in the egg or sperm cell at the time of fertilisation and can be passed down from parents to offspring. A pathogenic gene variant is one that is known to cause disease, such as conferring an increased risk of developing one or more types of cancer.

Some pathogenic gene variants increase risk of more than one cancer type. These gene variants can cause hereditary cancer ‘syndromes’ – i.e. collections of signs and symptoms with a common cause. The two most common hereditary cancer syndromes are:

* Hereditary Breast and Ovarian Cancer (HBOC) Syndrome - caused by a pathogenic BRCA1 or BRCA2 gene variant and characterised by increased risk for female and male breast cancer, ovarian cancer, prostate cancer, pancreatic cancer, melanoma and other cancers.
* Lynch syndrome, also called Hereditary Non-Polyposis Colon Cancer (HNPCC) - caused by a pathogenic gene variant of one of five genes – MLH1, MSH2, MSH6, PMS2 and EPCAM. Lynch syndrome is characterised by increased risk for colorectal, endometrial and other cancers.

The likely cancer risk for people with hereditary cancer syndromes varies by the gene variant (e.g. people with a BRCA1 variant have a different risk profile to those with a BRCA2 variant). It is important for patients and clinicians to know which variant an individual has in order to understand the associated risks and plan appropriate management.

## Indicators of possible cancer predisposition

The first step in harnessing the power of genetics in cancer risk reduction is to identify people who are at increased risk of having an inherited predisposition to cancer, and who should therefore be considered for genetic testing. For example, a suspicion of inherited cancer predisposition may be raised by a family history of cancers occurring at a young age, unusual types of cancer (e.g. male breast cancer), or patterns of cancers that suggest a hereditary cancer syndrome. Other clinical features or laboratory test results may indicate a likelihood, as outlined in figure 2 below.

**Figure 2 - Examples of clinical scenarios relating to hereditary cancer and the identification of a possible inherited cancer predisposition through genetic testing**

* **Individuals with a cancer diagnosis**
  + Genetic testing related to suitability for drug treatments
  + Features suggesting inherited risk, e.g. tumour pathology, many paediatric cancers, tumour genetic profiling carried out (e.g. in research context or prognostic)
  + Family history which suggests an inherited risk
* **Individuals without a cancer diagnosis/personal history of cancer**
  + Family history which suggests an inherited risk
  + Relative of a known carrier, i.e. cascade testing
  + Clinical syndrome or phenotype associated with a high risk of cancer (examples in both paediatrics and adult presentation)

## Genetics in cancer care and risk reduction

Genetics is now recognised as a growing and integral part of cancer care, with a role across treatment, early detection and risk reduction. Identification of cancer predisposition will only lead to patient benefit, if necessary services are in place and appropriately resourced and monitored. The potential benefit to a patient - and their relatives - can include reducing the risk of cancer developing, identifying cancer at an earlier more treatable stage, informing the optimal choice of targeted therapy if a cancer develops and facilitating reproductive options for the individual.

* **Risk reduction** – Genetics offers a direct and cost-effective way of identifying people who are at high risk of developing cancer. These people can then be managed appropriately, including offering interventions to reduce the risk of cancer developing, e.g. risk-reducing surgery and chemoprevention.
* **Early detection** – People who are identified as having an inherited cancer predisposition can be closely monitored to facilitate early detection and treatment of disease. Diagnosing cancer early typically allows for less complex treatment and optimal outcomes.

* **Treatment** - Certain cancer therapies rely on genetic and genomic information to tailor treatment to give the best chance of cure. For cancer patients with an inherited cancer predisposition, treatment decision-making based on timely genetic testing is rapidly becoming an international standard of care. Genetic test results guide surgical decisions and direct choice of medication. Targeting medication according to genetic test results (pharmacogenetics) is at the forefront of precision medicine.
* **Reproductive counselling and care** – Additional benefits of cancer genetics include the ability to identify people with a cancer predisposition and offer them appropriate counselling and care in relation to their reproductive health and options.
* **Benefit to relatives** – Cascade or predictive testing of unaffected relatives identifies more individuals with an inherited cancer predisposition, who can then benefit from the above cancer risk reduction and early detection measures.

The identification of cancer predisposition allows for the implementation of these risk-reduction measures, such as prophylactic surgery and chemoprevention, and surveillance to ensure early detection. In this way, genetic testing - for individuals and subsequent cascade testing of relatives - and provision of comprehensive downstream services can effectively reduce both cancer incidence and cancer mortality.

# Context

## Strategic context

### National Cancer Strategy 2017-2026

The National Cancer Strategy 2017-2026 highlighted the need for further development of hereditary cancer services in Ireland, with the aim of providing equitable access to services for all patients on a national basis. The strategy acknowledged the need for a strategic approach, to include increased infrastructural and financial support.

Relevant recommendations of the strategy include recommendations 6, 19 and 20:

**Recommendation 6** - The NCCP will draw up a plan by end-2017 for the development of an integrated cancer control and surveillance service for defined population subgroups with an inherited familial predisposition to cancer (e.g. breast, ovarian and colorectal).

**Recommendation 19** - The NCCP will further develop the Programme for Hereditary Cancers to ensure that evaluation, counselling, testing and risk reduction interventions are available as appropriate, and that services are available to patients on the basis of need.

**Recommendation 20** - The HSE will ensure that the existing cancer genetics services are amalgamated into one National Cancer Genetics Service and will identify the most appropriate site for its location.

Priorities highlighted in the strategy include cancer control and surveillance services for those with an inherited familial predisposition to breast, ovarian and colorectal cancer.

### National Genetics and Genomics Strategy

Development of genetics and genomics services in Ireland, to benefit all relevant clinical disciplines, is a current priority. In May 2022, the HSE constituted a National Genetic and Genomic Steering Group to guide the development of a National Genetic and Genomic Strategy for Ireland, due for publication in December 2022.

The strategy will also seek to address a number of core issues common to specialist genetics across multiple specialty areas. These include workforce challenges, in particular the need for a training and registration pathway for genetic counsellors in Ireland and recruitment and retention challenges across the services as a whole.

The Specialist Cancer Genetics Service will represent a core component of overall clinical genetics services in Ireland. It is envisaged that a new National Office for Genetics and Genomics, in collaboration with the NCCP, will provide Governance for the Specialist Cancer Genetics Service.

## Current hereditary cancer services in Ireland

At present, hereditary cancer services in Ireland are underdeveloped and under-resourced to meet the rapid growth in service demand *(National Cancer Strategy)*. This is not unique to cancer, hence the need identified for a National Genetic and Genomic Strategy for Ireland, to drive the necessary investment and development in genetics, to support a broad range of specialties.

Currently, specialist genetics services for hereditary cancer are provided in St James’s Hospital (SJH) Cancer Genetics Service (adults only) and at the Department of Clinical Genetics, Children's Health Ireland (CHI) at Crumlin (adult and paediatric). Referrals are received from both primary and secondary care. Services engage with patients via a range of in-person, virtual and telephone clinics and postal methods.

Services provided include:

* Genetic/family risk assessment, education and genetic counselling
* Germline testing
* Interpretation of results, including variants of uncertain significance
* Recommendations in relation to appropriate follow-up and ongoing management
* Cascade germline testing for relatives
* Reproductive options advice

Patients are typically discharged to the referring clinician, with follow-up recommendations. Given the absence of a structured system of referral for ongoing care, patients may be directed towards a known service in their geographical area or this may be left to the referring clinician to coordinate.

A number of established services exist, with considerable expertise in relation to certain cancer predisposition syndromes, e.g. specifically in relation to breast cancer predisposition, or colorectal cancer predisposition. However, these currently operate independently of each other and without an agreed pathway to/from specialist cancer genetics.

## Defining the requirements for hereditary cancer services in Ireland

Deficiencies and challenges exist across all elements of the hereditary cancer services in Ireland. These challenges are defined in the National Cancer Strategy 2017-2026, and in various other reports and assessments, including an environmental scan of the cancer genetics services published by the Irish Cancer Society in April 2021, and a health needs assessment for people with cancer predisposing BRCA gene variants, published by the National Cancer Control Programme in 2022 (Ref).

### Health needs assessment for persons diagnosed with a cancer-predisposing variant of BRCA1 and BRCA2 in Ireland

A health needs assessment published by the NCCP in 2022 highlighted the improvements required to ensure a high quality, comprehensive service for carriers of pathogenic BRCA gene variants. The needs assessment highlighted deficiencies across eight key themes, and made associated recommendations for service improvement (Appendix X). Many of the issues raised are challenges common to other inherited cancer predisposition syndromes.

* The need for accessible, inclusive and jargon-free information resources for carriers and information resources for health care professionals working in areas other than specialist cancer genetics
* A system required to gather data to support national planning and co-ordination of services for the affected population and facilitate local follow-up of patients
* Need for improved access to suitably qualified specialist genetics healthcare professionals after diagnosis, to include family planning supports and specialist fertility services
* A structured care pathway is required to ensure consistency in and access to optimal standards of care in all geographic areas and improved co-ordination of care
* Adequate access to risk-reducing measures, such as prophylactic surgery and surveillance, require dedicated resources, protected from the demands of the symptomatic service
* The need for access to psychological support throughout the patient journey and
* Specialist services relating to the adverse effects of risk-reducing measures, in this case the management of surgical menopause

### The unmet need in cancer genetic services: conducting an environmental scan of the cancer genetics service in an Irish context underpinned by a mixed methods approach - Report prepared for the Irish Cancer Society

This report provided an overview of current cancer genetic services in Ireland, highlighted deficiencies and made a suite of recommendations to optimise services (Table 1).

**Table 1: Recommendations of the ICS report**

|  |  |
| --- | --- |
| **Number** | **Recommendation** |
| **1** | Implement a hub and spoke model with genetics expertise within the dispersed oncology system. Genetics needs to be formally integrated into the cancer treatment pathway with uniform access to genetic testing, molecular tumour boards and access to genetics expertise and support at the point of care for both patients and their clinicians. |
| **2** | Build and further develop the genetics workforce and capability. |
| **3** | Increase cancer genetic diagnostics capability and expertise in Ireland. |
| **4** | Use a data management system that tracks referrals, appointments, and receipt of diagnosis with associated key performance indicators in terms of time to appointments, time to receipt of genetic test results and time to receipt of follow-up interventions (if required). |
| **5** | Streamline the genetics pathway to optimise online data collection and processing of data ensuring that follow-up counselling and health promoting interventions for individuals with pathogenic variants is optimised. |
| **6** | Increase knowledge and awareness of health care professionals, patients and the public of genetics and genetic services. |
| **7** | A dedicated pathway for individuals with specific syndromes or mutations with audited quality assured key performance indicators is required e.g. BRCA, Hereditary Breast and Ovarian Cancer Syndrome, Lynch Syndrome. Such pathways will ensure coordination of timely access to evidence-based surveillance, screening, surgery, and treatments as needed for individuals with specific mutations. |
| **8** | Test interventions that support the communication of information relating to genetic mutations with family members. |
| **9** | Explore and address the barriers to cascade testing of at-risk relatives. |
| **10** | Address concerns relating to the management of clinical samples and genetics data. |

## Summary - challenges facing hereditary cancer services in Ireland

As highlighted through these various needs assessments and reports, the challenges facing hereditary cancer services in Ireland include, but are not limited to, the following:

* There is no nationally agreed model of care for patients with inherited cancer predispositions.
* There are no nationally agreed guidelines for genetic assessment and testing in Ireland (a number of international guidance documents are followed, e.g. ERN[[1]](#footnote-1) Genturis, NICE Breast guidelines and UKCGG guidelines).
* Existing hereditary cancer services are under-developed and under resourced. There is a lack of capacity to deliver care across all elements of the pathway, including genetic testing, genetic counselling, surveillance, risk reducing surgery, related health needs (e.g. fertility/reproductive care) and follow-up care.
* Waiting lists for specialist cancer genetic services are extensive, and patients often have prolonged waits for their results. Thus, results with therapeutic relevance for patients undergoing treatment are delayed and healthy individuals are not being informed of their inherited cancer risk in a timely way.
* There is a lack of sufficient integration and coordination of care between existing specialist cancer genetic services, regional cancer services and primary care.
* There is inequitable access to existing services.
* Many healthcare professionals lack sufficient training/genetic literacy to advise and inform service users in relation to hereditary cancer services.
* There is a lack of appropriate data sources and IT infrastructure to support the planning, delivery, monitoring and improvement of hereditary cancer services, including the absence of a registry of patients with inherited cancer predispositions.

## Optimising hereditary cancer services in Ireland

### Hereditary cancer services model of care

This model of care defines the vision for an optimal Hereditary Cancer Service for the population of Ireland, guided by the needs identified above.

Implementation of the model will be a complex, multi-year project.

A core concept which will be included in the proposed framework is that of ‘mainstreaming’ genetics and how this can be implemented in a safe and efficient manner in cancer services in Ireland.

### The concept of mainstreaming genetics

Mainstreaming in cancer genetics refers to a model whereby the provision of cancer genetic testing for those with a personal cancer diagnosis is integrated into oncology services. This is an approach which is also being adopted across many other non-cancer specialties, as a means of increasing access to genetic testing for patients,

Mainstreamed pathways enable access to diagnostic genetic testing for patients with cancer as part of the standardised management of cancer care, in line with nationally agreed criteria and protocols.

A member of the team treating the patient, e.g. medical oncologist, surgeon or clinical nurse specialist, carries out the pre-test counselling, in accordance with agreed protocols and supported by necessary training. The identification of a pathogenic gene variant then triggers referral to specialist cancer genetics services for post-test counselling and further information in relation to cascade testing of family members.

Referral onward to specialist genetics is also required when a variant of uncertain significance (VUS) is identified, or where no variant is identified but a significant family history warrants genetics input.

A number of healthcare care institutions internationally have implemented such a model, likely in response to such factors as:

* Increased clinical utility of genetic testing, including in selection of appropriate drug treatment, necessitating quicker access to testing and results
* Improving genetic literacy among both the public and non-genetics healthcare professionals
* A recognition of the need to increase identification of those with inherited cancer predisposition where action can be taken to reduce cancer risk
* An inability worldwide of the specialist genetics workforce to meet this growing demand.

The literature on this area (see Appendix X for bibliography) focusses most commonly on mainstreaming of BRCA testing for those with a diagnosis of ovarian cancer or breast cancer. The Royal Marsden Hospital is widely regarded as a leader in this area, with many centres, particularly other centres in the UK, using the Royal Marsden materials to train staff as part of implementing mainstreaming in their services.

Overall, patient experience and feedback on pre-test counselling and testing within their oncology appointments has been positive (ref). Where compared to traditional clinical genetic service testing models, for most the mainstreamed approach did not result in any major differences in satisfaction, knowledge and/or distress.

Importantly, several studies report substantially reduced waiting times for genetic testing following implementation of a mainstreamed model, and a small number of studies reported cost-savings within their service.

One Australian study reported on the integration of a genetic counsellor into oncology clinics; however, this approach would be limited by existing staff resources in a given healthcare system and is much less feasible compared to the mainstreamed approach using oncology staff.

Overall, the majority of oncologists and clinical nurse specialists studied indicated positive feedback and acceptance of the mainstreamed approach. One UK study reported that most of the breast surgeons surveyed were not in favour of the mainstreamed approach due to existing constraints on their time and perceived lack of expertise, preferring to maintain their role in triaging patients by referral to clinical genetics.

Genetics professionals studied were overall supportive, although in one study less than 50% of the geneticists or genetic counsellors surveyed responded that patients seemed to receive accurate information about the BRCA test in the pre-test counselling session.

These findings highlight the potential of mainstreaming to improve patient care, if planned for and supported appropriately. Education and training of healthcare professionals involved in mainstreamed pathways appears to have positive impacts on confidence and knowledge of healthcare professionals to engage in pre-genetic test discussions with patients and address questions. Communication between non-genetics and genetics services in mainstreamed pathways has been highlighted as a priority, both in the pre-implementation and operational phases of these services. It is therefore key that a move towards a mainstreamed model for cancer genetics in Ireland would happen in conjunction with further development of the specialist cancer genetics services, to ensure a safe and supported mainstreamed service, in addition to the ongoing specialist cancer genetics role.

# Vision for the model of care

The vision of this model of care is that it will provide clarity regarding the structure and governance of hereditary cancer services in Ireland. The model of care will ensure/support:

* Development of national guidance to underpin standardised best practice, including mainstreaming of cancer genetic testing
* Equitable access to hereditary cancer services for all
* Clear governance and clarity regarding the roles of national genetics and genomic structures, specialist cancer genetics services and regional cancer predisposition services, within an overarching model of care
* Commitment to the Sláintecare principles of right person, right place, right time – through the integration of genetic assessment and testing into regional services, with access to national specialist genetic expertise as required
* Appropriate use of technology
* Timely access to genetics assessment and testing, to specialist cancer genetics expertise as required and to risk reduction options
* Comprehensive and coordinated approach to ongoing management
* Quality assurance of service
* Access to high quality patient information, psychological support and peer support for patients
* Education and training of health care professionals, including accredited training for those delivering mainstreamed pathways

## Evidence-based national guidance

Standards, protocols and guidelines should be agreed nationally, in accordance with best available evidence, best international practice, and as applied to the Irish context.

The full patient pathway should be addressed, from identification of those requiring family risk/ genetic assessment, use of assessment tools, criteria for genetic testing and types of testing employed (e.g. a national test directory similar to NHS) and recommendations on surveillance, follow up and risk reduction. Guidance should be easily available to clinicians, e.g. maintained online.

Guidance is specifically required to support the implementation of mainstreamed pathways. Mainstreamed pathways enable access to diagnostic genetic testing for patients with cancer as part of the standardised management of cancer care, in line with nationally agreed criteria and protocols.

## Equity of access

The service should ensure equitable access to optimal care for all, regardless of area of residence. This does not mean all aspects of care are best delivered locally. Certain services will be most appropriately delivered supraregionally/ nationally, for reasons of quality and efficiency, but equity of access must be assured for all.

## Clarity of roles within an overarching model of care

A clear governance framework is required which will outline the role of the new national genetics and genomic programme in relation to the agreement of national standards, practices and protocols and management guidelines; education and training; specialised services e.g. in relation to family planning and how this will operate in partnership with the NCCP in relation to cancer services.

Clarity is also required in relation to role and governance of both specialist genetics services and regional cancer predisposition services.

## Right person, right place, right time – assessment and testing

Cancer services should be upskilled to ensure ability to assess family history risk (in patients affected/ unaffected by cancer) and to implement mainstreamed testing pathways (related to drug treatment or other treatment indications).

For a person with a diagnosis of cancer, mainstreamed pathways ensure that genetic assessment (and testing where appropriate) is part of standard management. Post test counselling and management recommendations for those with a pathogenic variant should then be provided in line with national guidelines and delivered with specialist cancer genetics input as required. Input will also be required in relation to interpretation of variants of uncertain significance (VUS).

Where testing is considered in an unaffected individual (e.g. relative as part of cascade testing), that is most appropriately managed by specialist cancer genetics, i.e. a trained genetic counsellor. As regional access to specialist cancer genetics input improves, collaborative approaches to cascade testing can be developed which utilise the experience of a skilled regional staff member. A GP referral should not be required for relatives to avail of cascade testing.

Specialist cancer genetics services are also required for support for complex cases, either for remote advice, as a tertiary referral point +/- discussion at a national MDT.

## Appropriate use of technology

The use of technology, such as virtual assessment and decision aid tools, should be employed appropriately to maximise efficiency and support use of protocols.

Consideration should be given to patient preference, particularly if there is potential for receiving an initial diagnosis virtually, mindful of the potential for issues with digital literacy, healthcare literacy, language barriers, education level and poverty. Virtual appointments can be easier and less costly for patients. The patient can still attend a more local hospital for the virtual appointment.

Group genetic counselling is also possible and could improve efficiency.

## Timely access to genetics assessment +/- testing and result

As highlighted above, employment of mainstreaming pathways as appropriate will improve timely access to genetic testing for those with a cancer diagnosis.

For all patients, there should be an agreed timeframe for patients to be offered genetic testing and for a specialist cancer genetics consultation when required (before or after test result), to reduce anxiety and to allow for timely surveillance/ chemoprevention/ risk reducing surgery options to be offered/provided. A monitoring framework will be required.

The turnaround time for testing should facilitate timely treatment decisions and risk reduction measures, and be acceptable to patients.

## Timely access to risk-reduction options

A dedicated pathway with protected resources is required for management of those identified with a pathogenic variant/ inherited cancer predisposition – across prevention, surveillance, risk-reducing surgery, chemoprevention and wider supportive management (e.g. psychological support, management of adverse effects of risk reduction measures, reproduction and fertility). The most appropriate location for services will vary depending on the prevalence of the condition and the complexity of the intervention.

In particular, the appropriate configuration of services for risk-reducing surgery should be examined, to identify those that would result in low wait times and the best clinical outcomes for patients. Consideration should also be given to the specialist pathology input required.

A sufficiently resourced system, supported by necessary IT platform(s), is required to follow patients and ensure interventions can be put offered/delivered in a timely fashion at the appropriate age.

## Patient access to specialist genetics expertise

Those with an identified cancer predisposition/ pathogenic gene variant should have access to suitably qualified healthcare professionals after they receive their results, to ask any questions that arise after they have had a chance to process their diagnosis.

Some of these clinical queries could be addressed by designated genetics-trained healthcare professional(s) (e.g Advanced Nurse Practitioners (ANPs)) in cancer centres, while other queries, including reproductive counselling, would fall solely within the remit of specialist genetics services (e.g. Genetic Counsellors or genetic consultants).

Individuals with a pathogenic gene variant should be provided with information regarding specialist advice on reproduction and fertility. Fertility services in this context should be made available in the public sector and in a timely fashion.

## Comprehensive and coordinated approach to ongoing management

Improved coordination of care is required, including between specialist cancer genetics services and regional services/cancer centres. This can be supported via the agreement of a national model of care, including patient pathways.

Comprehensive services require access to wider supportive management (e.g. psychological support, management of adverse effects of risk reduction measures, reproduction and fertility), in addition to risk reduction measures.

Ongoing clinical care of those with an identified inherited cancer predisposition could be delivered by a nurse-led service, with the CNS/ANP also acting as the dedicated point of

contact. There is benefit to the autonomous clinical role of an ANP in this context.

The coordinated approach to management at regional level, including adherence to protocols and regular audit, should sit within an overarching framework for governance of clinical care of hereditary cancer services.

## Quality assurance of service

All elements of the patient care pathway (structures, processes and outcomes) should have defined quality standards and a subset of specific key performance indicators to facilitate performance measurement.

Patient-reported experience measures should be an embedded quality indicator.

## Access to high quality patient information

Accessible, inclusive (across age, language and ethnicity) and jargon-free informational resources should be developed for, and available as part of routine care, to those undergoing genetic testing and those identified as carriers.

Any such resources should be co-designed with patient stakeholders.

## Psychological support and peer support

Psychological support including professional support should be offered (but not mandatory) to all carriers following a diagnosis of a pathogenic variant. This should be available if needed throughout the patient journey, including but not only at times such as undergoing risk-reducing surgery and in the event of a cancer diagnosis.

Education initiatives should meet the education and training needs of the psychology profession specific to inherited cancer predisposition.

A pathway should be established for carriers to access psychological supports (both professional and peer-based) as requried, including those external to cancer centres.

## Education and training of health care professionals

Development of information resources specifically for healthcare professionals working in primary care and cancer services in Ireland is required. This should be informed by consultation with clinical subject matter experts.

Education of the cancer services workforce is required, to support implementation of guidelines, pathways and protocols, including accredited training for those delivering mainstreamed pathways.

# Service delivery/structure

## Three tier model of service delivery

A programmatic approach is required to ensure national agreement of guidance, including mainstreaming protocols, laboratory and clinical data infrastructure, specialist genetics workforce development, and education of the wider workforce. This programme will be a collaboration between by the National Genetics and Genomics Office and the National Cancer Control Programme.

Delivery of hereditary cancer services will be organised across three ‘tiers’:

* Generalist level
* Regional Cancer Predisposition Services
* Specialist Cancer Genetics Services

This model of care outlines roles of each tier and the working relationships to be established between the ‘tiers’, e.g. the way in which the Specialist Cancer Genetics Service will support and enable delivery of care by Regional Cancer Predisposition Services. It is essential that patient care across the tiers is seamless, with clear communication and coordination of care.

**Figure 3: Hereditary cancer services - model of service delivery**



## Generalist Services

Healthcare services (including primary care and non-oncology services in secondary care) should be empowered to recognise those with a possible inherited predisposition to cancer and refer appropriately and to understand the management of those with identified cancer predisposition.

Referral pathways and protocols will be agreed nationally, and specific education initiatives should focus on those working in generalist services, to ensure appropriate knowledge of referral pathways/protocols and identification of patients for referral. The need for improved genetic literacy across all healthcare professionals is recognised in the draft National Genetics and Genomics Strategy.

## Regional Cancer Predisposition Services

Regional services have a role in the identification and assessment of those with a possible inherited predisposition to cancer, including [mainstreamed diagnostic testing](#_The_concept_of) for inherited pathogenic gene variants in patients with cancer, according to national protocols.

Regional services also deliver the necessary healthcare to those identified with an inherited cancer predisposition, to determine role for targeted cancer therapy where relevant, to reduce cancer risk and/or facilitate early detection and to maximise patient health and wellbeing.

Multiple services including medical oncology, surgical oncology, haematology and benign services such as gastroenterology are involved in the identification and management of cancer predisposition.

The required multi-disciplinary service for people with the more common cancer predisposition syndromes (e.g. BRCA carriers, Lynch syndrome) should be coordinated regionally at cancer centre level and requires a structure to facilitate follow-up of patients. Certain rarer syndromes may follow a different model, with patient management and follow-up delivered in one centre, in close collaboration with the specialist cancer genetics service.

Regional cancer predisposition services require designated input from specialist cancer genetics services, with a clear clinical governance structure.

### **Regional Cancer Predisposition Services - Workforce**

#### **Clinical Lead Role**

A **Clinical Lead for Cancer Predisposition** should be appointed with an identified sessional commitment to lead the development of each Regional Cancer Predisposition Service. This role will include leading:

* development of regional service (tailored to population size and specialisms)
* adherence to national protocols
* development of local pathways
* ensuring appropriate education and training of local workforce. All cancer services should be upskilled to ensure the provision of a high-quality service capable of accurately assessing family history risk (in patients affected/ unaffected by cancer) and to effectively implement mainstreamed testing pathways (related to drug treatment or other indications for treatment).
* clinical audit & service improvement
* service monitoring, including measuring and reporting KPIs

***Requirement:*** *Each regional service will require an identified clinical lead for this role, e.g. with a sessional commitment equivalent to 0.4WTE. A lesser time commitment may be feasible in smaller centres, or if sharing this role across two sites within a network/ future Regional Health Area.*

#### **Administrative support**

Appropriate administrative support will be integral to service delivery, monitoring and evaluation. These roles should include data management and clinic coordination. Genetics and genomics has a higher requirement for record retrieval and documentation than other specialties, due e.g. to the detailed information provided by patients on their family history, or the tracking of multiple pathology test results. **Genomics Resource Associates** will play an essential non-clinical support role in this area, including support for patients in navigating the assessment and testing process and the subsequent management.

Technological solutions may alleviate some of administrative burden associated with service delivery.

#### **Genetic Counsellors**

The role of **Genetic Counsellor** is a Specialist Cancer Genetics role, but will be delivered regionally. It will be possible for a Genetic Counsellor to be either appointed and employed locally by the Regional Cancer Predisposition Service, or centrally by the Specialist Cancer Genetics Service. Where appointed regionally, a professional reporting relationship to and sessional commitment to Specialist Cancer Genetics will be required.

A Genetic Counsellor will fulfil the following roles within each of the Regional Cancer Predisposition Services:

* act as point of contact for support for non-geneticists who are providing pre-test counselling
* develop and deliver educational initiatives to the wider workforce
* provide comprehensive pre or post-test genetic counselling as required, including reproductive advice/information
* act as point of contact/link to the Specialist Cancer Genetics tertiary service for complex cases and consultation on variants of unknown significance
* contribute to research

***Requirement:*** *Each of the regional cancer predisposition services requires an identified genetic counsellor, appointed either locally or centrally, with a professional reporting relationship to a Principal Genetic Counsellor in the specialist cancer genetics service.*

*This will not equate to a WTE cancer genetics counsellor per cancer centre, as a sessional commitment will be included to the national hub/network, including national MDTs.*

*Smaller cancer centres may choose to operate as part of a network (/future Regional Health Area) with another cancer centre for this service.*

#### **Specialty Lead**

Within specific specialties (e.g. medical oncology, colorectal, gynaecology, breast), a **lead consultant or ANP** should lead on implementing Regional Cancer Predisposition Services, including ensuring access to mainstreamed testing where applicable and ongoing management of those with an identified cancer predisposition.

The exact configuration of specialties and their role in delivery of Regional Cancer Predisposition Services will vary according to services available in the region and may change over time – e.g. breast, gynaeoncology, colorectal, upper GI, medical oncology, haematology. Collaborative working will be key, as patient needs frequently cross clinical specialties.

#### **Nursing**

CNS and ANP members of teams are key to the delivery of the Regional Cancer Predisposition Services, with roles including assessment of family history, patient pre-test counselling and provision of continuity of care and coordinated care for those identified with a cancer predisposition. Administrative support in this area will allow skilled nursing staff to work at the top of their licence and will support the coordination of care across different clinical areas.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Sample staff involvement in Regional Cancer Predisposition Service** | | | | | |
|  | | | | **Clinical Lead** |  |
| **Medical Oncology** | **Colorectal**  **service** | **Gynaecology** | **Breast service** |  | **Genetic Counsellor\*** |
| *Consultant* | *Consultant* | *Consultant* | *Consultant* | Admin staff |  |
| *CNS* | *CNS* | *CNS* | *CNS* | Genetics Resource Associate | |
| *ANP* | *ANP* | *ANP* | *ANP* |  |  |
| *Pathology, radiology, endoscopy, psychooncology services* | | | | | |

\*may be employed by cancer centre or specialist cancer genetics service but professional reporting relationship to specialist cancer genetics

## Specialist Cancer Genetics Service

The Specialist Cancer Genetics Service will function as the national centre for clinical expertise in Ireland and will provide support and advice to Regional Cancer Predisposition Service sites via a network of designated genetic counsellors, a tertiary referral service and national MDT(s).

The role of the Specialist Cancer Genetics Service will include:

* develop national guidance, including protocols for mainstreamed services as appropriate. All standards, protocols and guidelines will be agreed nationally, and founded on the best available evidence and best international practice, appropriately applied in the Irish context.
* horizon scanning - as cancer genetics and genomic services and techniques continue to advance, it is important to ensure that the introduction of new tests and phasing out of others is informed by specific guidelines, and that this will be audited;
* develop and deliver educational initiatives to the wider workforce. Guidance is specifically required to support the implementation of mainstreamed pathways.
* support non-geneticists who are providing pre-test counselling
* provide a regional predictive genetic testing service
* provide comprehensive pre or post-test genetic counselling as required, including reproductive advice/information
* provide a tertiary referral service for complex cases and consultation on variants of unknown significance (VUS) – referral pathways will be agreed to facilitate referral of patients to the Specialist Cancer Genetics Service by a clinical team based at the Regional Cancer Predisposition Services.
* Support in the management of complex cases may be provided remotely or in person at a tertiary referral centre (+/- discussion at a national MDT).

Adult and paediatric patients have differing needs and should be assessed and managed in an appropriate environment (see below).

### **Specialist Cancer Genetics Services - Workforce**

**Consultants**

Consultant expertise in specialist cancer genetics is provided by (i) those trained as Consultants in Clinical Genetics and (ii) those trained as oncology consultants, who have developed expertise in the area of cancer genetics, specifically cancer predisposition. Those trained in oncology have a greater understanding of the overlap with current or future cancer treatments, whereas clinical geneticists have a broader role in areas such as syndrome overlap with non-cancer areas. Both areas of expertise are valued and should be maintained.

Of note, the oncology consultant role referred to here is different to that defined within Regional Cancer Predisposition Services. The lead role regionally may be held by someone with an interest in service development in this area but would not require the clinical expertise to provide a tertiary referral specialist cancer genetics service. However, regional expertise can be expected to develop further over time, particularly in the area of more common hereditary cancer syndromes.

**Genetic Counsellors**

As previously outlined, some genetic counsellors will provide a dedicated service to regional cancer predisposition services but will maintain both a sessional commitment and a professional reporting relationship to the specialist cancer genetics service.

Genetic Counsellors will also operate as part of the tertiary referral service in cancer specialist genetics, will have a role in education, training and guidance development and provide a clinical governance structure for genetic counsellors working regionally.

Challenges in Ireland have been noted in relation to lack of career pathway and registration for genetic counsellors. This has been highlighted as a priority issue for the national genetics and genomics office.

**Administrative and IT support**

As outlined for regional services, the administrative workload for genetics is high compared to other specialties. The team will also benefit from Genetic Resources Associates and support to ensure coordination of national MDT(s) and the genetic counsellor network.

**Laboratory expertise**

Molecular diagnostic expertise is required as part of the specialist cancer genetics MDT, particularly in relation to the interpretation of Variants of Uncertain Significance.

## Paediatric and AYA Cancer Predisposition Service

Childhood cancer, including that in adolescents/young adulthood, is a rare but complex disease, with differing cancers and different disease biologies to those seen in adults. In addition, these cancers are associated with a higher rate of cancer predisposition, with less influence of environmental factors. Currently, it is estimated that up to 30% of young patients with childhood or adolescent/young adult cancers (CAYA) require genetic evaluation, with at least 10% having a confirmed cancer predisposition syndrome.

A comprehensive service is therefore required that incorporates cancer predisposition risk assessment for all CAYA patients. In addition, as standard of care is moving internationally towards somatic testing of all tumours diagnosed in the CAYA population, germline testing for all will likely be required, either in sequence or parallel, necessitating expertise in relation to interpretation and management.

Children’s cancer services are already centralised at the CHI cancer centre (currently in Crumlin) and the development of a formal national network for CAYA services is currently underway (reference). The provision of a single national specialist paediatric MDT for cancer genetics fits with this model and such a service has already commenced at CHI at Crumlin. The multidisciplinary input required crosses specialist genetics, oncology/haematology, molecular pathology and the wider CAYA cancer team. Collaboration will be required between this MDT and that at the AYA service in St James’ Hospital, to meet the needs of this age group.

In addition to providing access from regional paediatrics services to/from this specialist service, consideration should also be given to cross over with regional cancer predisposition services who are dealing with adults. For example, it may be appropriate for genetic testing to be carried out in children of adults diagnosed with certain cancer predisposition syndromes. These children should be referred to specialist genetics services associated with paediatric cancer predisposition services, for age-appropriate genetic counselling and management. The benefit of consultant clinical geneticists and genetic counsellors on the MDT is that they are skilled to deal with all ages and all types of clinical presentations (syndromic/non-syndromic), cancer risks and risks of other medical conditions, across all age spectrums.

Similarly, when cancer predisposition is identified in a child, testing of adults may identify those with the same predisposition and who will benefit from ongoing management in their regional cancer predisposition service.

Specific challenges can arise in relation to disease surveillance in this population, particularly younger children. These include greater reliance on U/S and MRI techniques, as minimising use of radiation, and the need for a general anaesthetic for certain surveillance procedures.

# Clinical Pathways

Detailed clinical pathways will need to be developed, or adopted from international guidelines, in relation to a broad range of possible cancer predisposition.

Patient pathways should include the following:

* identification of those requiring family risk/ genetic assessment
* details of assessment tools to be used
* formal criteria for genetic testing
* types of testing to be employed (e.g. a national test directory similar to NHS) and
* recommendations on follow-up and risk reduction.

Such pathways and guidance will be agreed under the governance of the National Genetics and Genomics Office (once established) and the National Cancer Control Programme and should be easily available to clinicians, e.g. maintained online.

Generic pathways are presented here to outline the proposed patient flow across regional cancer predisposition services and specialist cancer genetics.

## Pathways for referral, risk assessment and testing of people who may have a cancer predisposition

### **People with a cancer diagnosis**

The implementation of “mainstreamed” pathways as appropriate, will ensure timely access to genetic testing for those with a cancer diagnosis. A national timeline to provide patients with genetic consultations, and testing (if required) will need to be developed and agreed upon as part of a standard management programme. This is key to effectively managing patient anxiety and allowing for timely surveillance/ chemoprevention/risk-reducing surgery options to be explored.

## Pathway A: Adult Patients with a Cancer Diagnosis



### **People without a cancer diagnosis**

Where testing is undertaken in an individual who does not have a cancer diagnosis (e.g. a relative as part of cascade testing), this will be managed by a trained Genetic Counsellor.

Obtaining a GP referral should not be a barrier to relatives availing of cascade testing. However, a relative should be asked to provide GP details, so that their GP can receive information on outcome of testing and any follow-up recommendations. Referral for ongoing management and follow up, if required, should be to the relevant regional cancer predisposition service.

## 

## Pathway B: Patients without a personal history of cancer, other than cascade testing



## Pathway C: Cascade testing[[2]](#footnote-2) for unaffected relatives of those with an identified cancer predisposition gene



## Clinical pathways for management of people with identified cancer predisposition

A dedicated pathway with protected resources is required for the management of those identified with a pathogenic variant/ inherited cancer predisposition – across prevention, surveillance, risk-reducing surgery, chemoprevention and wider supportive management (e.g. psychological support, management of adverse effects of risk reduction measures, reproduction and fertility). This will be required, whether or not the patient has an existing diagnosis of cancer.

Pathway D outlines generic components of care for those with an identified cancer predisposition. Condition-specific pathways will be need to be agreed, including consideration of international best practice.

### **Key components of care for people diagnosed with a cancer predisposing genetic variant**

**Access to specialist cancer genetics services/expertise**

Those with a newly identified cancer predisposition/ pathogenic gene variant should have access to a suitably qualified healthcare professional after they receive their diagnosis, to ask any questions that arise after they have had a chance to process their diagnosis. A designated genetics-trained healthcare professional, e.g Advanced Nurse Practitioners (ANPs) in the Regional Cancer Predisposition Service/cancer centres, could address some of these clinical queries. Other requirements will fall within the remit of Specialist Cancer Genetic Services , e.g. Genetic Counsellors (including familial and reproductive counselling), although this specialist service may be delivered at regional level (model of care recommends access to a designated specialist genetic counsellor in each regional cancer predisposition service).

**Timely access to risk-reducing interventions**

A system is required for surveillance and follow up of patients to ensure interventions, including prophylactic surgery and chemoprevention, can be offered/delivered in a timely fashion at the appropriate age.

In particular, the appropriate configuration of services for risk-reducing surgery must be addressed, to minimise wait times and ensure the best clinical outcomes for patients. Condition-specific pathways must also address the need for specialist pathology input, when dealing with specimens from prophylactic surgeries in those with a known pathogenic gene variant.

**Psycho-oncology**

Psychological support (including professional and peer-based) should be offered (but not mandatory) to all carriers following a diagnosis of a pathogenic variant. This should be available if needed throughout the patient journey, including but not only at times such as pre/post risk-reducing surgery and in the event of a cancer diagnosis.

Additional education initiatives will be required to meet the training needs of the psychology profession, in relation to the area of inherited cancer predisposition.

**Reproduction and Fertility**

Carriers of pathogenic gene variants should be provided with information regarding specialist advice on reproduction and fertility. Fertility services in this context should be made available free at point of care in the public sector and in a timely fashion.

Regardless of the original pathway to identification, a comprehensive service is then required for those with a pathogenic/ likely pathogenic variant (Pathway D)

## Pathway D: Management Algorithm for those with a pathogenic/ likely pathogenic cancer predisposing genetic variant



# Key enablers of service delivery

Key enablers of effective delivery of cancer genetics services in Ireland include:

* Sufficient resources and funding to meet current and future service demand
* Clearly defined patient care pathways
* Governance
* Quality assurance, service monitoring and evaluation
* Appropriate IT infrastructure and data management
* Appropriate use of technology
* Education

## Sufficient resources and funding

Guaranteed long-term funding is required for the Specialist Cancer Genetics Service and the Regional Cancer Predisposition Services in order to meet current and future demand.

The services should be staffed by sufficient numbers of appropriately trained staff with clearly defined roles and responsibilities.

## Governance

A clear governance framework is required for hereditary cancer services in Ireland.

A National Genetics and Genomics Office is soon to be established, in line with the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland. It will have responsibility for agreement of national standards (practices and protocols), guidelines, education and training, and delivery of specialised genetic services e.g. family planning, and workforce development.

The National Cancer Control Programme should be closely engaged with in relation to the development of services related to an inherited predisposition to cancer and a process agreed for the development and agreement of guidance and pathways relevant to cancer predisposition.

The Specialist Cancer Genetics Services will have clinical responsibility for development and agreement of national guidance for their speciality and have professional oversight of specialist genetics services (such as genetic counselling) delivered within Regional Cancer Predisposition Services.

Within the Regional Cancer Predisposition Services, the Clinical Lead for Cancer Predisposition will ensure that staff are appropriately trained for and support in the tasks they are undertaking and that as a service, there is

* A coordinated approach to service management
* National protocols are adhered to and
* Regular audits are undertaken

## Quality assurance, service monitoring and evaluation

All elements of the patient care pathway (structures, processes and outcomes) should have defined quality standards and a subset of specific key performance indicators to facilitate service monitoring, quality assurance and service improvement.

Patient-reported experience measures should be embedded as a quality indicator. Experience of patients who have received/are receiving care from the Specialist Cancer Genetics Service and/or Regional Cancer Predisposition Services should be included in the cancer patients experience survey (Recommendation 35 of the National Cancer Strategy - The NCCP will define focused cancer patient experience surveys to incorporate treatment and survivorship in line with HIQA’s standard approach for the National In-Patient Acute Care Patient Experience Survey).

Audits should be undertaken to ensure equitable access to services irrespective of patients’ age, geographic location and socioeconomic status.

## Appropriate IT Infrastructure and data management

A well-integrated IT system, linked to all GP services and hospitals is required to enable effective service delivery.

A national database for inherited cancer predisposition, based on the use of a unique patient identifier, would support national planning and coordination of services for the population. Moreover, the development of a national database would allow health information to be accessed as required across different sites. Ideally, this would include data from public and private clinical services, while acknowledging that the HSE only has jurisdiction over/responsibility for publicly delivered health services in Ireland. Commercial solutions with integrated patient support packages should be considered.

The approach taken to IT infrastructure will need to align with that taken by the National Genetics and Genomics Office, re wider services, laboratory test results and patient registries.

Options include:

* Development of a national database for Specialist Cancer Genetics and Regional Cancer Predisposition Services
* Each Regional Cancer Predisposition Site to host an individual system.

Genetic and genomic information (test results, as opposed to raw data) should be an integral component of clinical data available to patients with a cancer diagnosis. A formal process will be required to record patient consent digitally, including the sharing of genetic information with family members.

At a minimum, an agreed minimum dataset should be established, to accurately record data on patients with a pathogenic gene variant. This would support Regional Cancer Predisposition Services to coordinate patient follow-up and actively participate in audit.

Genetic and genomic information (test results, as opposed to raw data) should be an integral component of clinical data available for an individual with a cancer diagnosis.

Each Regional Cancer Predisposition Service/site will require a data manager to facilitate reporting on service activity, and KPI reporting.

## Appropriate use of technology

The use of technology, such as virtual assessment and decision aid tools, should be employed appropriately to maximise efficiency and support the use of protocols.

Consideration should be given to equity of access (barriers may include digital poverty or insufficient digital literacy to enable virtual engagement with the services), and to patient preference, particularly if there is potential for receiving an initial diagnosis virtually. Virtual appointments can be more convenient and less financially costly for patients. Group genetic counselling is also possible, and could improve efficiency.

## Education

### Education of the workforce

Information resources and education initiatives are required for healthcare professionals working in primary care, non-oncology secondary care and cancer services in Ireland. These will support the implementation of guidelines, pathways and protocols, and should include accredited training for those delivering mainstreamed pathways (currently under development). These resources should be developed in consultation with clinical subject matter experts. Established genetic counselling training through existing academic support programmes.

### Patient Education

Patient stakeholders will be integral partners in the co-design of patient-facing educational resources.

# Implementation

The implementation of this Model of Care will be a multi-year project. An implementation group will be established to oversee this work, encompassing the following areas:

* Guidance and clinical pathway development (BRCA, Lynch, other)
* Workforce
* Healthcare professional education and training
* Data requirements and Quality Assurance
* Patient Information and supports (general, and specific to cancer predisposition syndromes, e.g. BRCA, Lynch syndrome etc.)
* Synergies with the National Genetics and Genomics Office, once established.

Recommendations for implementation are outlined here, for detailed population and refinement by the implementation steering group once established.

|  |
| --- |
| Thematic Area 1: |
| Establish oversight for the implementation of the Hereditary Cancer Model of Care |
| 1.1 Establish a governance and reporting structure aligned to the National Cancer Strategy and National Cancer Control Programme |
| 1.2 Agree areas for collaboration with the National Genetics and Genomics Office once in place |
| 1.3 Convene a multi-disciplinary implementation steering group, with clinical leadership and patient involvement |
| 1.4 Establish relevant working groups for thematic priorities |
| 1.5 Agree an overarching governance framework for hereditary cancer service |
| 1.6 Appoint Clinical Leads for Cancer Predisposition within regional services |
| 1.6 Report at minimum annually on progress in implementation |
| Thematic Area 2 : |
| Development of networked services and MDTs |
| 2.1 Define the process(es) by which genetic counselling services can be networked and made accessible regionally |
| 2.2 Develop a standardised pathway to a national specialist cancer genetics MDT (adults) |
| 2.3 Develop a standardised pathway to a national specialist cancer genetics MDT (CAYA) |
| 2.4 Establish a network of regional cancer predisposition leads |
| Thematic Area 3 : |
| Healthcare professional education and training; workforce development |
| 3.1 Identify the training needs of staff working in generalist settings |
| 3.2 Identify the training needs of staff working in regional cancer predisposition services |
| 3.3 Develop an accredited online programme to meet the minimal learning needs staff working in regional services |
| 3.4 Identify or develop further training courses to meet identified needs of staff |
| 3.5 Work with the National Genetics and Genomics Office, professional training bodies and regulators to develop career pathways in specialist cancer genetics |
| Thematic Area 4: |
| Clinical guidance and pathway development |
| 4.1 Agree the guidance development and approval process, in line with HSE policy, including guidance on laboratory testing |
| 4.2 Establish guidance development groups for common presentations and cancer predispositions |
| 4.3 Ensure patients are involved in the co-design of clinical pathways |
| 4.4 Agree the process by which international guidance relating to rarer conditions will be approved and disseminated for use in Ireland |
| 4.5 Agree the platform(s) through which current guidance will be made available to all |
| 4.5 Highlight critical points in clinical pathways, which should be subject to regular audit |
| 4.6 Describe resource requirements for implementation of pathways |
| Thematic Area 5: |
| Patient Information and supports |
| 5.1 Establish subgroups for development of patient information, according to common presentations and cancer predispositions |
| 5.2 Ensure patients are engaged in the co-design of patient information materials |
| 5.3 Consider how best to meet the information needs of underserved populations and those with health literacy challenges |
| 5.4 Agree or develop platform(s) through which patient information can be most easily accessed |
| 5.5 Ensure evaluation of information materials and supports provided |
| Thematic Area 6: |
| Data Requirements and IT infrastructure |
| 6.1 Explore the role of virtual patient assessment within hereditary cancer services |
| 6.2 Identify the IT infrastructure required to enable the implementation of this model of care, including a genetic counselling network and referral to a national specialist genetics MDT. |
| 6.3 Examine potential data capture options with the National Cancer Information System |
| 6.4 Engage with the National Genetics and Genomics Office in relation to development of registries relation to cancer predisposition |
| 6.5 Agree minimum datasets for common presentations and cancer predispositions |
| 6.6 Agree key performance indicators for the delivery of specialist cancer genetics and regional cancer predisposition services |

# Benchmarks of success

|  |  |  |
| --- | --- | --- |
| Strategic Priorities | | Details |
|  | Partnerships/MDT working | A networked approach to all public services treating patients undergoing cancer genetic testing across the state  Development of a formal network, encompassing the Specialised Cancer Genetics Service and Regional Cancer Predisposition Services  Timely access to genetics assessment +/- testing and result  Each cancer centre should have a clear pathway of how they refer patients with an identified cancer predisposition/ pathogenic gene variant(s) to suitably qualified healthcare professionals  Development of National MDMs  Creation of new services specific to the needs of patients within the Specialist Genetics Service and the Cancer Predisposition Service  Co-design of services by those with lived experience of the Specialist Genetics Service and the Regional Cancer Predisposition Service  A systematic and collaborative approach to the private and charity sector supporting patients receiving care in the Specialist Genetics Service and the Regional Cancer Predisposition Service  Representation by patients from the Specialist Genetics Service / the Regional Cancer Predisposition Service with lived experience contributing to relevant working groups within the NCCP  Representation by parent/family members in key working groups |
|  | Education and Information | The provision of accessible, jargon-free and inclusive/equitable (across age, language and ethnicity) educational resources  Development of additional education and training resources to improve genetic literacy among healthcare professionals  Provide healthcare professionals responsible for implementing mainstreamed pathways  with specific, accredited training |
|  | Workforce | An integrated network of healthcare professionals with special education and training in both the Specialist Cancer Genetics Service and Regional Cancer Predisposition Service  All Regional Cancer Predisposition Services should have an identified, appropriately skilled workforce, to include consultants, ANPs, CNSs and administrative support, and have dedicated access to the specialist genetics services.  Create, promote and nurture a network of skilled and highly motivated professionals across the spectrum of health and social care professions that are educated and skilled in identifying and addressing the specific and unique needs of service users within the Specialist Cancer Genetics Service and the Regional Cancer Predisposition Services.  Partner with the voluntary sector to create specific support for those receiving care from both the Specialist Genetics Service and Regional Cancer Predisposition Services |
|  | Research | Strengthen the research community within the Specialist Cancer Genetics Service and the Regional Cancer Predisposition Service in Ireland  Increase participation in multi-site international research to further develop evidence-based cancer genetics practice/services |
|  | Data | An ability to collect standardised data nationally to support planning, service monitoring and quality improvement  Creation of Key Performance Indicators and benchmarking against international standards/services |

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# Appendix B: Membership of Advisory Group

# Appendix C: BRCA Health Needs Assessment Recommendations

**Table C.1: Recommendations arising from the NCCP BRCA health needs assessment (Ref)**

|  |  |  |
| --- | --- | --- |
|  | **Theme** | **Recommendations** |
| **1** | Information needs | * 1. Accessible, inclusive (across age, language and ethnicity) and jargon-free informational resources should be developed for and available as part of routine care to BRCA carriers. Consultation with BRCA carrier stakeholders should be part of the development of any such resources.   2. Development of BRCA information resources specifically for healthcare professionals working in primary care and cancer centres in Ireland is required. This should be informed by consultation with clinical subject matter experts. |
| **2** | Data on BRCA in Ireland | * 1. A national database with an agreed minimum dataset should be established capturing data pertaining to BRCA carriers under follow-up in this country. Ideally this would include data from public and private clinical services. This could be developed as part of a national database for inherited cancer predisposition. Such a database should be based on the use of a unique patient identifier. It should support national planning and co-ordination of services for the BRCA population and facilitate local follow-up of patients. There should be adequate resources to maintain it.   2. Local databases, standardised with respect to an agreed minimum dataset i.e. a single data dictionary, should be established in cancer centres which capture data pertaining to BRCA carriers under follow-up. |
| **3** | Specialist Genetics input | 3.1 BRCA carriers should have access to suitably qualified healthcare professionals after their consultation at diagnosis with clinical genetics, to ask any questions that arise after they have had a chance to process their diagnosis. Some of these queries could be addressed by designated genetics-trained healthcare professional(s) (e.g Advanced Nurse Practitioners (ANPs) once appropriate training provided) in cancer centres, while other queries would fall within the remit of Genetic Counsellors.   * 1. BRCA carriers should be provided with information regarding, and signposted to, family planning resources including accessing pre-implantation genetic testing (PGD) and in-vitro fertilisation (IVF).   2. BRCA carriers should be considered eligible for publicly available onco-fertility services |
| **4** | Structured Care Pathway and Co-ordination of Care | * 1. Improved coordination of care is required, including between clinical genetics services and cancer centres. This could be delivered by a nurse-led service, with the CNS/ANP also acting as the dedicated point of contact for BRCA carriers.   2. Development of a nurse-led service for BRCA carriers will require specialist training and support. There is benefit to the autonomous clinical role of an ANP in this context.   3. The model of care (including clinical governance) for BRCA carriers needs to be improved and standardised across all cancer centres, to ensure consistency in and access to optimal standards of care in all geographic areas. Further dedicated consultation on the desired model of care is needed with input from all relevant stakeholders.   4. All elements of the patient care pathway (structures, processes and outcomes) should have defined quality standards and a subset of specific key performance indicators to facilitate performance measurement.   5. Patient-reported experience measures should be an embedded quality indicator. |
| **5** | Risk-reducing surgery | 5.1 - Risk-reducing breast and ovarian surgery for BRCA carriers requires a dedicated pathway with protected resources and should be delivered in a timely fashion. The appropriate configuration of services for risk-reducing surgery, that would result in low wait times and the best clinical outcomes for women, should now be examined.   * 1. Adequate post-operative support following risk-reducing surgery (e.g. physiotherapy, psychological support) should be available to patients.   2. All women should have timely access to specialist plastic surgery expertise regarding breast reconstruction options, and reconstructive surgery itself, if desired. |
| **6** | Surveillance | * 1. There should be protected magnetic resonance imaging (MRI) and mammography slots for breast surveillance of asymptomatic BRCA carriers, to ensure surveillance imaging occurs at recommended intervals   2. Ovarian surveillance by transvaginal ultrasound and/or Cancer Antigen (CA)-125 measurement is not recommended for BRCA carriers.   3. Female BRCA carriers who request ovarian surveillance should be made aware of the lack of evidence to demonstrate a survival benefit.   4. Greater awareness of the risks and benefits of prostate cancer surveillance for male BRCA carriers – particularly for BRCA2 – is needed among patients as well as healthcare professionals. It is not yet known whether surveillance using PSA reduces mortality in men with a cancer-predisposing BRCA variant   5. An evidence review of international pancreatic cancer surveillance guidelines for BRCA carriers is needed. |
| **7** | Psychological support | * 1. Psychological support including professional support should be offered (but not mandatory) to all BRCA carriers following a diagnosis of a cancer-predisposing BRCA variant. This should be publicly available if needed throughout the patient journey, including but not only at times such as undergoing risk-reducing surgery and in the event of a cancer diagnosis.   2. Education initiatives should include the education and training needs of the psychology profession specific to inherited cancer predisposition.   3. BRCA carriers should be signposted following diagnosis to psychological supports (both professional and peer-based), including those external to cancer centres. |
| **8** | Women’s health | * 1. Women should have access to clear information regarding the symptoms and longer-term health risks associated with early menopause, prior to risk-reducing surgery.   2. Women should have access to expert advice regarding their options for menopause management (hormonal and non-hormonal), which could be provided by a suitably trained doctor, ANP or other adequately trained healthcare professional.   3. Women should be referred to specialist menopause clinics if adequate support or expertise is not available in their hospital. |

1. European Reference Network [↑](#footnote-ref-1)
2. Also known as predictive testing [↑](#footnote-ref-2)